Conus Medullaris Spinal Arteriovenous Malformation in a Patient with Klippel-Trenaunay-Weber Syndrome

A Case Report and Review of the Literature

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Summary

We describe a 24-year-old woman who presented with twice previously unverified subarachnoid hemorrhages from the conus medullaris spinal arteriovenous malformation with Parkes-Weber-syndrome.

Spinal MRI examination is considered to be necessary for the diagnosis of Klippel-Trenaunay-Weber syndrome. For diagnosis of the spinal cord arteriovenous malformation, it is indispensable to search carefully for the presence of accompanying lesions. Transarterial glue embolization is effective for the management of the spinal vascular lesion.

Introduction

Klippel-Trenaunay syndrome (KTS) is a neurocutaneous syndrome mainly characterized by hypertrophy of unilateral lower or upper limbs with cutaneous hemangioma and vascular malformations in the affected limb. This syndrome is rarely associated with a spinal cord arteriovenous malformation. Since Den Hartog Jegar's report [1], 26 cases of this rare association have been published [2–18]. This paper reports a case of Parkes-Weber syndrome (PWS) which is a variant of the KTS with conus medullaris spinal arteriovenous malformation, developed by subarachnoid hemorrhage on two occasions. The focus of this report is the interest of MRI in delineating associated cord lesions in Klippel-Trenaunay or Parkes-Weber syndromes by the use of embryological links.

Case Report

A 24-year-old woman was admitted to the department of neurosurgery because of the sudden loss of consciousness during dinner. The patient was diagnosed with subarachnoid hemorrhage. The MRA and cerebral angiography could not identify the source of the hemorrhage. The patient underwent a V-P shunt for hydrocephalus and was discharged after one month hospitalization.

Another five months later she complained of a headache at home and was readmitted to the same hospital because of the diagnosis of a subarachnoid hemorrhage.

The computed tomography (CT), magnetic resonance image (MRI), magnetic resonance angiography (MRA), and cerebral angiography revealed no obvious lesion of the hemorrhage. Although cerebral infarction, caused by vasospasm in right parietal lobe appeared, only

Abbreviations: KTS = Klippel-Trenaunay syndrome, PWS = Parkes-Weber syndrome, AVMs = arteriovenous malformations, AVFs = arteriovenous fistulas, MRI = magnetic resonance imaging, MRA = magnetic resonance angiography, CT = computed tomography.

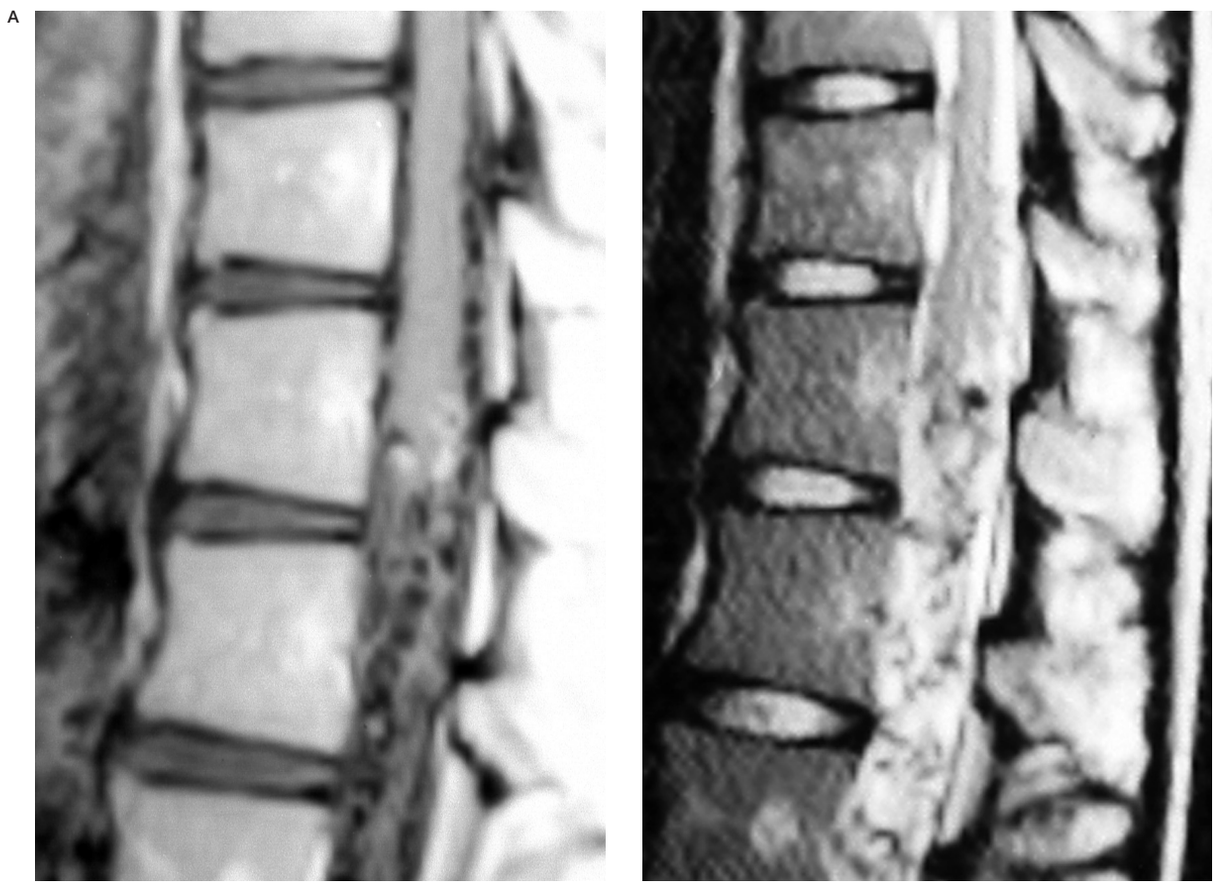

Figure 1 A) MRI T1WI with Gd in sagittal view demonstrates gadolinium enhanced vascular lesions around conus medullaris. B) MRI T2WI in sagittal view demonstrates vascular flow void around conus medullaris.

conservative treatment resulted in the improvement of the symptom. The patient then left three weeks after hospitalization.

The patient visited the department of neurology in our hospital. Flow void was observed in the subarachnoid space around the conus medullaris on the spinal MRI (figure 1A,B). Spinal arteriovenous malformation was strongly suspected.

Therefore, she was referred to the department of radiology to investigate the cause of the subarachnoid hemorrhage, and be closely examined for spinal vascular disease. Spinal angiography revealed a conus medullaris arteriovenous malformation composed of a total of four feeding arteries around the conus medullaris; pial spinal artery from the left second lumbar artery (figure 2A), an anterior spinal artery from the left ninth intercostal artery (figure 2B), and posterior lateral spinal arteries from the right tenth intercostal artery and the left eleventh intercostal artery (not shown). Since the patient displayed hyperplasia of left lower extremity (figure 3), and port-wine stain hemangioma and arteriovenous fistula with varicose vein of left lower limb (figure 4A-D), the patient was diagnosed with conus medullaris spinal arteriovenous malformation in Parkes-Weber syndrome.

To prevent recurrent bleeding caused by the spinal arteriovenous malformation, endovascular intervention was performed under general anesthesia. Embolization with a microcatheter (Magic FM 0.012 Balt Co. Paris) was performed through the second lumbar artery (figure 2C) to reduce the nidus volume, selectively using NBCA (figure 2D). There were no complications by transcatheter embolization (figure 2E). Capillary vascular malformation around the left ankle and foot and arteriovenous fistula in the left lower limb developed ulcer formation and were treated surgically by a plastic

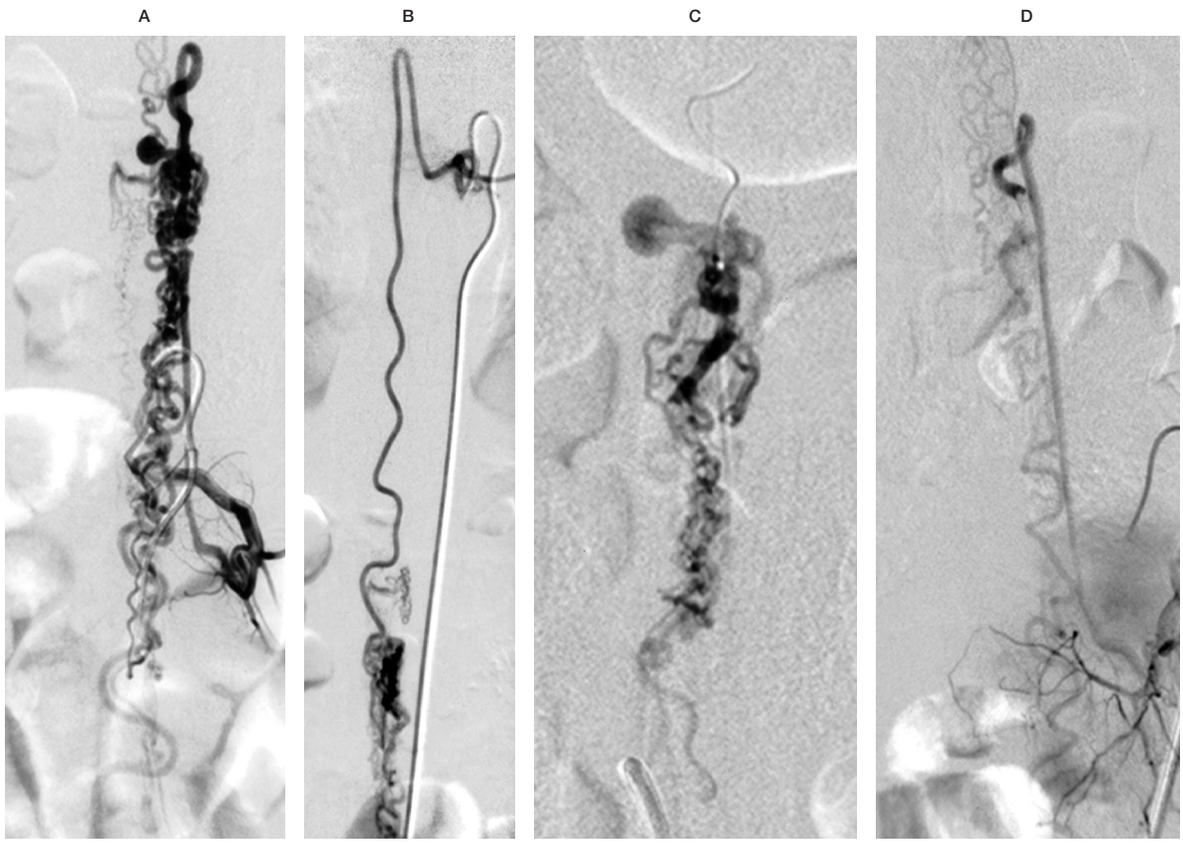

Figure 2 A) Radiculopial artery arising from the left L2 is the main feeder to the malformation around the conus medullaris harboring an intranidal aneurysm. B) Other contributors came from the anterior spinal artery arising from the left 9th. intercostalis artery demonstrates the anterior component of spinal arteriovenous malformation. C) Super selective approach of arteriovenous malformation performed through the left L2 radiculopial artery. D) Glue embolization to the nidus. E) Immediate control angiogram through the same route shows a good disconnection of the nidus with respect to the parent vessel.

surgeon. As for hypertrophy in the left lower limb, we are following this patient without any active treatment.

There is no re-bleeding from the conus medullaris spinal arteriovenous malformation during a six year follow-up period.

Discussion

Klippel-Trenaunay syndrome was identified by Trenaunay (France) in 1900, and has been mainly characterized by the triad of varicous veins, cutaneous capillary vascular malformation and hypertrophy of all the tissues of the involved limb [19]. Parkes Weber then reported a similar case with arteriovenous fistula as a part of the syndrome [20].

The name of Klippel-Trenaunay-Weber (KTW) syndrome was proposed by Mullins et Al. to include these two syndrome and is well accepted because it is not always easy to differentiate arteriovenous fistula from varicose vein and some typical Klippel-Trenaunay syndromes also have arteriovenous fistula on angiography [21].

Twenty-six cases of Klippel-Trenaunay-Weber syndrome with spinal arteriovenous malformation have been reported. Five cases of them are not reported detail in individual cases. The symptoms consisted of six cases of subarachnoid hemorrhage and 15 cases of ischemia. Djindjian et Al reported an exceptional case of an 18-year-old woman with typical syndromes developed in her upper limb while spinal arteriovenous malformation was distributed in thoracic spine [4-5].

In all the other 20 cases, arteriovenous malformation developed at the level of the inferior part of the spinal cord while the typical syndromes were found in the lower limb [3]. Thoracic spine 4 was the highest level of spinal arteriovenous malformation; a 49-year-old woman

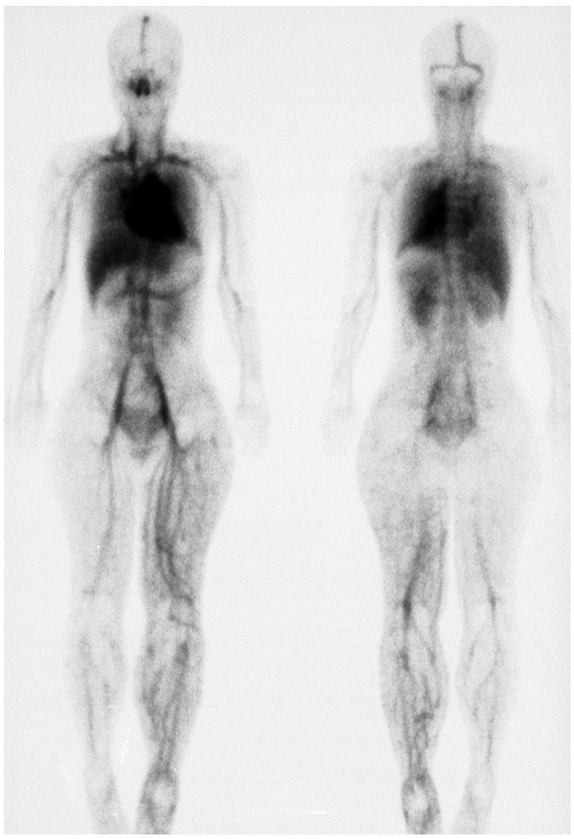

Figure 3 Hemihypertrophy of the left leg as well as a capillary hemangioma around the left ankle by radioisotope scintigram.

reported by Saito et Al[14], and a 47-year-old man reported by Fukutake et Al[6]. In these cases, spinal cord vascular lesions were found in the associated lesion with limb vascular malformations, such as in our present case. Twenty eight per cent of patients with spinal vascular malformations presented associated vascular malformations or dysplasias in previous reports. Sixteen per cent of them presented associated metameric lesions.

This group is always multiple, sharing potential metameric links. These lesions are multiple shunts for the metameric disposition cannot always be clearly demonstrated but can be postulated.

The angioarchtecture in the metameric group is suggested with a high possibility of the capillary arteriovenous shunt states[18]. Although the etiology of this peripheral angiopathy in KTWS is not clear, there were some close links between the spinal AVFs and the limb AVFs in previous KWT series. One must pay attention to search for the spinal vascular malformation thoroughly by MRI in a patient with limb hypertrophy and limb vascular malformation.

These spinal cord arteriovenous shunts are not related to a hereditary disorder; they are genetic but nonhereditary. Alvoet et Al investigated the genetics in 86 patients and found two familial cases[22]. Ceballos-Quintal et Al also de-

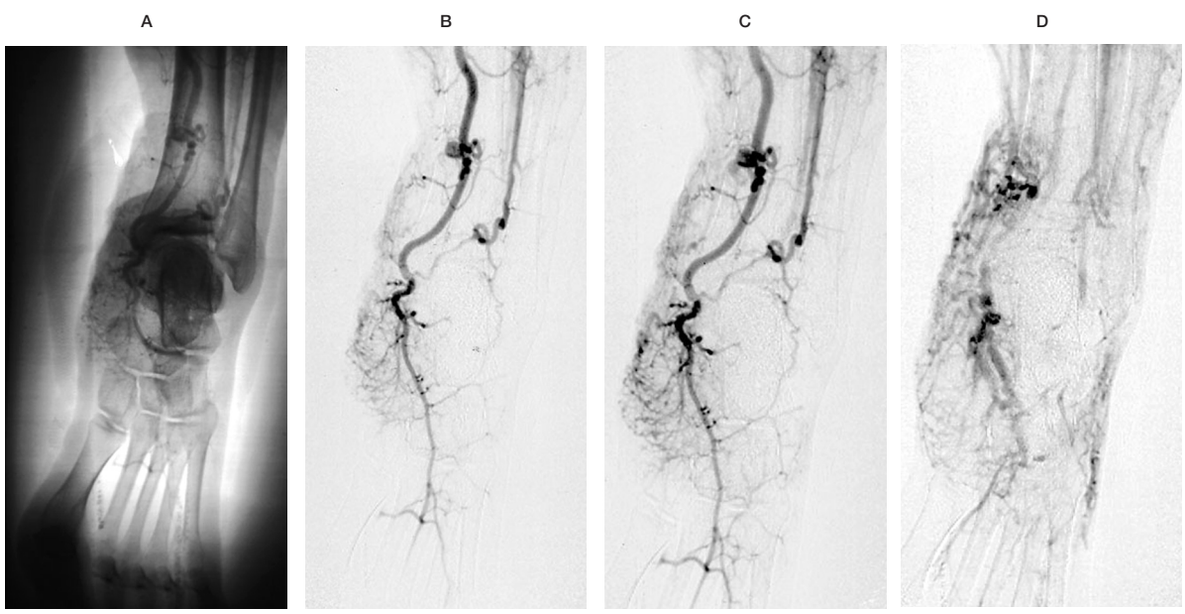

Figure 4 A) Left ankle ray-sum image. B) Left anterior tibial arteriography in arterial phase demonstrate the arteriovenous fistulae with varicose vein. C,D) Left anterior tibial arteriography in capillary and venous phases demonstrate the hemangioma around left ankle joint fed by tarsal arteries.

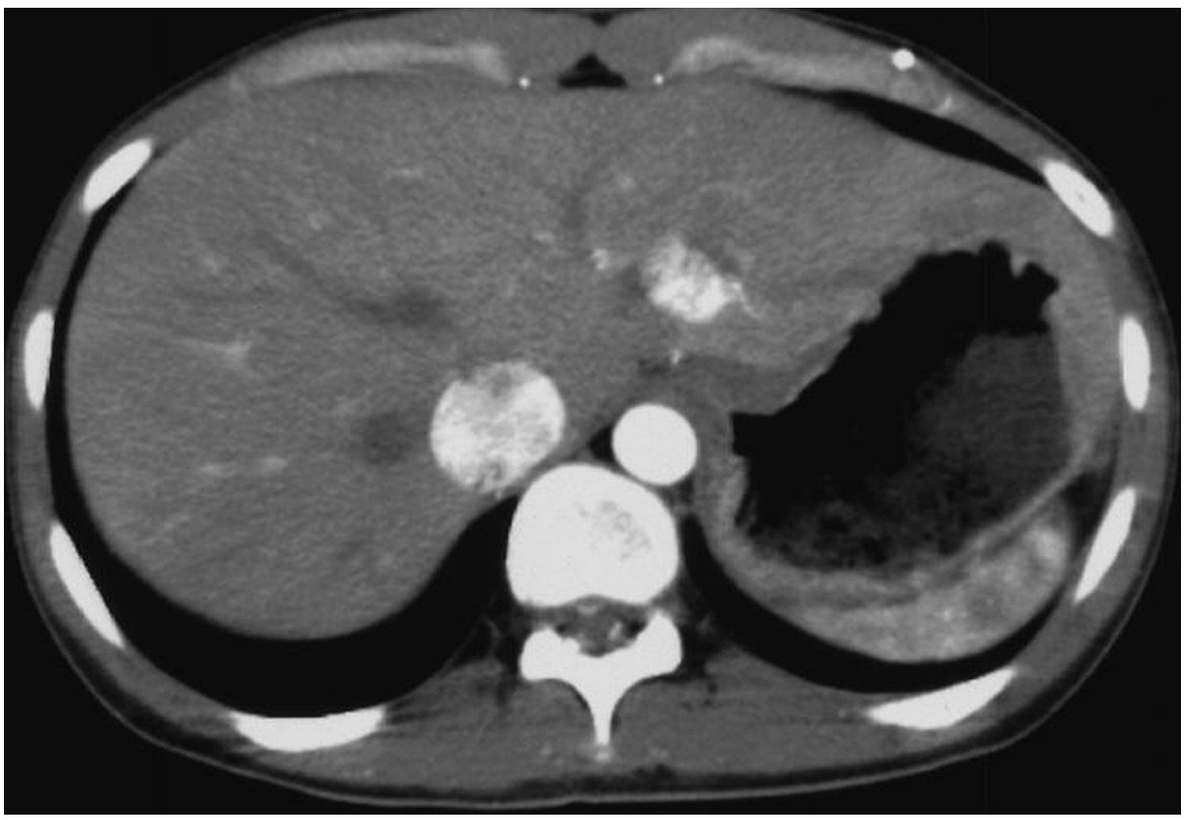

Figure 5 Abdominal computed tomography with contrast administration demonstrated hemangioma in left liver lobe.

scribed one familial case, whose mother had severe varicosities and a large capillary hemangioma on her back, and whose grandmother had developed varicositis at a young age [23]. Although some familial cases have been described, in general Klippel-Trenaunay syndrome is sporadic in appearance.

Our patient has no apparent familial history. The genetic analysis for KWT performed by Whelan et Al revealed that a single gene defect of chromosomes, human, Pair 5 (5p) or Pair 11(11p) caused metastasis in the proportion of five parts to 11 (a reciprocal translocation [46,XX, t (5; 11) (q13.3; p15.1)] [24]. Much work still has to be done to understand the molecular mechanisms of these rare anomalies. In case of the suspicion of KWT, genetic analysis should also be performed routinely

The treatment and follow-up observation after intervention in KWT patients should focus on the spinal vascular lesion as well as the associated lesions. Both neuroradiological imaging and genetic analysis will be essential diagnostic methods in future for these spinal vascular malformations.

Conclusions

We described a patient who had Parkes-Weber syndrome with spinal arteriovenous malformation.

When the accompanying lesions such as hypertrophy of unilateral lower or upper limb with cutaneous hemangioma and arteriovenous fistulas in the affected limb are diagnosed, it is indispensable to search for a spinal cord AV malformation by MRI. Our present case impressed that the spinal MRI examination is considered to be necessary for the diagnosis of Klippel-Trenaunay-Weber syndrome.

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